

TECH CENTER 1600/2900

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NOV 05 2001 #8

## RAW SEQUENCE LISTING

PATENT APPLICATION: US/09/597,732

DATE: 10/30/2001

TIME: 16:54:41

Input Set : N:\Crf3\RULE60\09597732.txt

Output Set: N:\CRF3\10302001\I597732.raw

3 <110> APPLICANT: Keating, Mark T.  
 4 Sanguinetti, Michael C.  
 5 Curran, Mark E.  
 6 Landes, Gregory M.  
 7 Connors, Timothy D.  
 8 Burn, Timothy C.  
 9 Splawski, Igor

11 <120> TITLE OF INVENTION: KVLQT1 - A LONG QT SYNDROME GENE  
 13 <130> FILE REFERENCE: 2323-133  
 15 <140> CURRENT APPLICATION NUMBER: 09/597,732  
 16 <141> CURRENT FILING DATE: 2000-06-19  
 18 <150> PRIOR APPLICATION NUMBER: 09/135,010  
 19 <151> PRIOR FILING DATE: 1998-08-17  
 21 <150> PRIOR APPLICATION NUMBER: 60/094,477  
 22 <151> PRIOR FILING DATE: 1998-07-29  
 24 <150> PRIOR APPLICATION NUMBER: 08/921,068  
 25 <151> PRIOR FILING DATE: 1997-08-29  
 27 <150> PRIOR APPLICATION NUMBER: 08/739,383  
 28 <151> PRIOR FILING DATE: 1996-10-29  
 30 <150> PRIOR APPLICATION NUMBER: 60/019,014  
 31 <151> PRIOR FILING DATE: 1995-12-22  
 33 <160> NUMBER OF SEQ ID NOS: 116  
 35 <170> SOFTWARE: PatentIn Ver. 2.0  
 37 <210> SEQ ID NO: 1  
 38 <211> LENGTH: 3181  
 39 <212> TYPE: DNA  
 40 <213> ORGANISM: Homo sapiens  
 42 <220> FEATURE:  
 43 <221> NAME/KEY: CDS  
 44 <222> LOCATION: (163)..(2190)  
 46 <400> SEQUENCE: 1

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 49 gggctggcag cagtggctgc ccgcaactgcg cccggggcgt cgccttcgct gcagctcccg 120  
 51 gtgcgcgcgc tggggccggc cccccggcag gccctcctcg tt atg gcc gcg gcc 174  
 52 Met Ala Ala Ala  
 53 1

55 tcc tcc ccg ccc agg gcc gag agg aag cgc tgg ggt tgg ggc cgc ctg 222  
 56 Ser Ser Pro Pro Arg Ala Glu Arg Lys Arg Trp Gly Trp Gly Arg Leu  
 57 5 10 15 20

59 cca ggc gcc cgg cgg ggc agc gcg ggc ctg gcc aag aag tgc ccc ttc 270  
 60 Pro Gly Ala Arg Arg Gly Ser Ala Gly Leu Ala Lys Lys Cys Pro Phe  
 61 25 30 35

63 tcg ctg gag ctg gcg gag ggc ggc ccg gcg ggc ggc gcg ctc tac gcg 318  
 64 Ser Leu Glu Leu Ala Glu Gly Gly Pro Ala Gly Gly Ala Leu Tyr Ala  
 65 40 45 50

67 ccc atc gcg ccc ggc gcc cca ggt ccc gcg ccc cct gcg tcc ccg gcc 366  
 68 Pro Ile Ala Pro Gly Ala Pro Gly Pro Ala Pro Pro Ala Ser Pro Ala

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69	55	60	65	
71	gcg ccc gcc gcg ccc cca gtt gcc tcc gac ctt ggc ccg cgg ccg ccg	414		
72	Ala Pro Ala Ala Pro Pro Val Ala Ser Asp Leu Gly Pro Arg Pro Pro			
73	70 75 80			
75	gtg agc cta gac ccg cgc gtc tcc atc tac agc acg cgc cgc ccg gtg	462		
76	Val Ser Leu Asp Pro Arg Val Ser Ile Tyr Ser Thr Arg Arg Pro Val			
77	85 90 95 100			
79	ttg gcg cgc acc cac gtc cag ggc cgc gtc tac aac ttc ctc gag cgt	510		
80	Leu Ala Arg Thr His Val Gln Gly Arg Val Tyr Asn Phe Leu Glu Arg			
81	105 110 115			
83	ccc acc ggc tgg aaa tgc ttc gtt tac cac ttc gcc gtc ttc ctc atc	558		
84	Pro Thr Gly Trp Lys Cys Phe Val Tyr His Phe Ala Val Phe Leu Ile			
85	120 125 130			
87	gtc ctg gtc tgc ctc atc ttc agc gtg ctg tcc acc atc gag cag tat	606		
88	Val Leu Val Cys Leu Ile Phe Ser Val Leu Ser Thr Ile Glu Gln Tyr			
89	135 140 145			
91	gcc gcc ctg gcc acg ggg act ctc ttc tgg atg gag atc gtg ctg gtg	654		
92	Ala Ala Leu Ala Thr Gly Thr Leu Phe Trp Met Glu Ile Val Leu Val			
93	150 155 160			
95	gtg ttc ttc ggg acg gag tac gtg gtc cgc ctc tgg tcc gcc ggc tgc	702		
96	Val Phe Phe Gly Thr Glu Tyr Val Val Arg Leu Trp Ser Ala Gly Cys			
97	165 170 175 180			
99	cgc agc aag tac gtg ggc ctc tgg ggg cgg ctg cgc ttt gcc cgg aag	750		
100	Arg Ser Lys Tyr Val Gly Leu Trp Gly Arg Leu Arg Phe Ala Arg Lys			
101	185 190 195			
103	ccc att tcc atc atc gac ctc atc gtg gtc gtg gcc tcc atg gtg gtc	798		
104	Pro Ile Ser Ile Ile Asp Leu Ile Val Val Ala Ser Met Val Val			
105	200 205 210			
107	ctc tgc gtg ggc tcc aag ggg cag gtg ttt gcc acg tgc gcc atc agg	846		
108	Leu Cys Val Gly Ser Lys Gly Gln Val Phe Ala Thr Ser Ala Ile Arg			
109	215 220 225			
111	ggc atc cgc ttc ctg cag atc ctg agg atg cta cac gtc gac cgc cag	894		
112	Gly Ile Arg Phe Leu Gln Ile Leu Arg Met Leu His Val Asp Arg Gln			
113	230 235 240			
115	gga ggc acc tgg agg ctc ctg ggc tcc gtg gtc ttc atc cac cgc cag	942		
116	Gly Gly Thr Trp Arg Leu Leu Gly Ser Val Val Phe Ile His Arg Gln			
117	245 250 255 260			
119	gag ctg ata acc acc ctg tac atc ggc ttc ctg ggc ctc atc ttc tcc	990		
120	Glu Leu Ile Thr Thr Leu Tyr Ile Gly Phe Leu Gly Leu Ile Phe Ser			
121	265 270 275			
123	tgc tac ttt gtg tac ctg gct gag aag gac gcg gtg aac gag tca ggc	1038		
124	Ser Tyr Phe Val Tyr Leu Ala Glu Lys Asp Ala Val Asn Glu Ser Gly			
125	280 285 290			
127	cgc gtg gag ttc ggc agc tac gca gat gcg ctg tgg tgg ggg gtg gtc	1086		
128	Arg Val Glu Phe Gly Ser Tyr Ala Asp Ala Leu Trp Trp Gly Val Val			
129	295 300 305			
131	aca gtc acc acc atc ggc tat ggg gac aag gtg ccc cag acg tgg gtc	1134		
132	Thr Val Thr Thr Ile Gly Tyr Gly Asp Lys Val Pro Gln Thr Trp Val			
133	310 315 320			

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135	ggg	aag	acc	atc	gcc	tcc	tgc	ttc	tct	gtc	ttt	gcc	atc	tcc	ttc	ttt	1182
136	Gly	Lys	Thr	Ile	Ala	Ser	Cys	Phe	Ser	Val	Phe	Ala	Ile	Ser	Phe	Phe	
137	325					330					335					340	
139	gcg	ctc	cca	gcg	ggg	att	ctt	ggc	tcg	ggg	ttt	gcc	ctg	aag	gtg	cag	1230
140	Ala	Leu	Pro	Ala	Gly	Ile	Leu	Gly	Ser	Gly	Phe	Ala	Leu	Lys	Val	Gln	
141					345					350						355	
143	cag	aag	cag	agg	cag	aag	cac	ttc	aac	cgg	cag	atc	ccg	gcg	gca	gcc	1278
144	Gln	Lys	Gln	Arg	Gln	Lys	His	Phe	Asn	Arg	Gln	Ile	Pro	Ala	Ala	Ala	
145				360					365					370			
147	tca	ctc	att	cag	acc	gca	tgg	agg	tgc	tat	gct	gcc	gag	aac	ccc	gac	1326
148	Ser	Leu	Ile	Gln	Thr	Ala	Trp	Arg	Cys	Tyr	Ala	Ala	Glu	Asn	Pro	Asp	
149			375					380					385				
151	tcc	tcc	acc	tgg	aag	atc	tac	atc	cgg	aag	gcc	ccc	cgg	agc	cac	act	1374
152	Ser	Ser	Thr	Trp	Lys	Ile	Tyr	Ile	Arg	Lys	Ala	Pro	Arg	Ser	His	Thr	
153			390				395					400					
155	ctg	ctg	tca	ccc	agc	ccc	aaa	ccc	aag	aag	tct	gtg	gtg	gta	aag	aaa	1422
156	Leu	Leu	Ser	Pro	Ser	Pro	Lys	Pro	Lys	Lys	Ser	Val	Val	Val	Lys	Lys	
157	405					410					415					420	
159	aaa	aag	ttc	aag	ctg	gac	aaa	gac	aat	ggg	gtg	act	cct	gga	gag	aag	1470
160	Lys	Lys	Phe	Lys	Leu	Asp	Lys	Asp	Asn	Gly	Val	Thr	Pro	Gly	Glu	Lys	
161				425						430					435		
163	atg	ctc	aca	gtc	ccc	cat	atc	acg	tgc	gac	ccc	cca	gaa	gag	cgg	cgg	1518
164	Met	Leu	Thr	Val	Pro	His	Ile	Thr	Cys	Asp	Pro	Pro	Glu	Glu	Arg	Arg	
165			440						445					450			
167	ctg	gac	cac	ttc	tct	gtc	gac	ggc	tat	gac	agt	tct	gta	agg	aag	agc	1566
168	Leu	Asp	His	Phe	Ser	Val	Asp	Gly	Tyr	Asp	Ser	Ser	Val	Arg	Lys	Ser	
169			455						460					465			
171	cca	aca	ctg	ctg	gaa	gtg	agc	atg	ccc	cat	ttc	atg	aga	acc	aac	agc	1614
172	Pro	Thr	Leu	Leu	Glu	Val	Ser	Met	Pro	His	Phe	Met	Arg	Thr	Asn	Ser	
173		470					475						480				
175	ttc	gcc	gag	gac	ctg	gac	ctg	gaa	ggg	gag	act	ctg	ctg	aca	ccc	atc	1662
176	Phe	Ala	Glu	Asp	Leu	Asp	Leu	Glu	Gly	Glu	Thr	Leu	Leu	Thr	Pro	Ile	
177	485				490						495					500	
179	acc	cac	atc	tca	cag	ctg	cgg	gaa	cac	cat	cgg	gcc	acc	att	aag	gtc	1710
180	Thr	His	Ile	Ser	Gln	Leu	Arg	Glu	His	His	Arg	Ala	Thr	Ile	Lys	Val	
181				505						510					515		
183	att	cga	cgc	atg	cag	tac	ttt	gtg	gcc	aag	aag	aaa	ttc	cag	caa	gcg	1758
184	Ile	Arg	Arg	Met	Gln	Tyr	Phe	Val	Ala	Lys	Lys	Lys	Phe	Gln	Gln	Ala	
185				520					525					530			
187	cgg	aag	cct	tac	gat	gtg	cgg	gac	gtc	att	gag	cag	tac	tcg	cag	ggc	1806
188	Arg	Lys	Pro	Tyr	Asp	Val	Arg	Asp	Val	Ile	Glu	Gln	Tyr	Ser	Gln	Gly	
189			535					540						545			
191	cac	ctc	aac	ctc	atg	gtg	cgc	atc	aag	gag	ctg	cag	agg	agg	ctg	gac	1854
192	His	Leu	Asn	Leu	Met	Val	Arg	Ile	Lys	Glu	Leu	Gln	Arg	Arg	Leu	Asp	
193		550					555							560			
195	cag	tcc	att	ggg	aag	ccc	tca	ctg	ttc	atc	tcc	gtc	tca	gaa	aag	agc	1902
196	Gln	Ser	Ile	Gly	Lys	Pro	Ser	Leu	Phe	Ile	Ser	Val	Ser	Glu	Lys	Ser	
197	565					570					575					580	
199	aag	gat	cgc	ggc	agc	aac	acg	atc	ggc	gcc	cgc	ctg	aac	cga	gta	gaa	1950

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200 Lys Asp Arg Gly Ser Asn Thr Ile Gly Ala Arg Leu Asn Arg Val Glu
201          585          590          595
203 gac aag gtg acg cag ctg gac cag agg ctg gca ctc atc acc gac atg 1998
204 Asp Lys Val Thr Gln Leu Asp Gln Arg Leu Ala Leu Ile Thr Asp Met
205          600          605          610
207 ctt cac cag ctg ctc tcc ttg cac ggt ggc agc acc ccc ggc agc ggc 2046
208 Leu His Gln Leu Leu Ser Leu His Gly Gly Ser Thr Pro Gly Ser Gly
209          615          620          625
211 ggc ccc ccc aga gag ggc ggg gcc cac atc acc cag ccc tgc ggc agt 2094
212 Gly Pro Pro Arg Glu Gly Gly Ala His Ile Thr Gln Pro Cys Gly Ser
213          630          635          640
215 ggc ggc tcc gtc gac cct gag ctc ttc ctg ccc agc aac acc ctg ccc 2142
216 Gly Gly Ser Val Asp Pro Glu Leu Phe Leu Pro Ser Asn Thr Leu Pro
217 645          650          655          660
219 acc tac gag cag ctg acc gtg ccc agg agg ggc ccc gat gag ggg tcc 2190
220 Thr Tyr Glu Gln Leu Thr Val Pro Arg Arg Gly Pro Asp Glu Gly Ser
221          665          670          675
223 tgaggagggg atggggctgg gggatgggcc tgagtgagag gggaggccaa gaggggcccc 2250
225 acctggccct ctctgaagga ggccacctcc taaaaggccc agagagaaga gcccactct 2310
227 cagaggcccc aatacccat ggaccatgct gtctggcaca gcctgcactt gggggctcag 2370
229 caaggccacc tcttcttggc cgggtgtggg gccccgtctc aggtctgagt tgttaccca 2430
231 agcgccctgg ccccccacat gtgatgttga catcactggc atggtggttg ggaccagtg 2490
233 gcagggcaca gggcctggcc catgtatggc caggaaagtag cacaggctga gtgcaggccc 2550
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247 gagccactg tgcgtggggc tcccgcctcc aaccctcgc ccagtcccag cagccagcca 2970
249 aacacacaga aggggactgc cactccctc tgccagctgc tgagccgcag agaagtgcg 3030
251 gttcctacac aggacagggg ttcttcttgg gcattacatc gcatagaaat caataatttg 3090
253 tgggtgatttg gatctgtgtt ttaatgagtt tcacagtgtg attttgatta ttaattgtgc 3150
255 aagcttttcc taataaacgt ggagaatcac a 3181
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259 <211> LENGTH: 676
260 <212> TYPE: PRT
261 <213> ORGANISM: Homo sapiens
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265 1          5          10          15
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268          20          25          30
270 Lys Cys Pro Phe Ser Leu Glu Leu Ala Glu Gly Gly Pro Ala Gly Gly
271          35          40          45
273 Ala Leu Tyr Ala Pro Ile Ala Pro Gly Ala Pro Gly Pro Ala Pro Pro
274          50          55          60
276 Ala Ser Pro Ala Ala Pro Ala Ala Pro Pro Val Ala Ser Asp Leu Gly
277 65          70          75          80

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```

279 Pro Arg Pro Pro Val Ser Leu Asp Pro Arg Val Ser Ile Tyr Ser Thr
280      85      90      95
282 Arg Arg Pro Val Leu Ala Arg Thr His Val Gln Gly Arg Val Tyr Asn
283      100      105      110
285 Phe Leu Glu Arg Pro Thr Gly Trp Lys Cys Phe Val Tyr His Phe Ala
286      115      120      125
288 Val Phe Leu Ile Val Leu Val Cys Leu Ile Phe Ser Val Leu Ser Thr
289      130      135      140
291 Ile Glu Gln Tyr Ala Ala Leu Ala Thr Gly Thr Leu Phe Trp Met Glu
292 145      150      155      160
294 Ile Val Leu Val Val Phe Phe Gly Thr Glu Tyr Val Val Arg Leu Trp
295      165      170      175
297 Ser Ala Gly Cys Arg Ser Lys Tyr Val Gly Leu Trp Gly Arg Leu Arg
298      180      185      190
300 Phe Ala Arg Lys Pro Ile Ser Ile Ile Asp Leu Ile Val Val Val Ala
301      195      200      205
303 Ser Met Val Val Leu Cys Val Gly Ser Lys Gly Gln Val Phe Ala Thr
304      210      215      220
306 Ser Ala Ile Arg Gly Ile Arg Phe Leu Gln Ile Leu Arg Met Leu His
307 225      230      235      240
309 Val Asp Arg Gln Gly Gly Thr Trp Arg Leu Leu Gly Ser Val Val Phe
310      245      250      255
312 Ile His Arg Gln Glu Leu Ile Thr Thr Leu Tyr Ile Gly Phe Leu Gly
313      260      265      270
315 Leu Ile Phe Ser Ser Tyr Phe Val Tyr Leu Ala Glu Lys Asp Ala Val
316      275      280      285
318 Asn Glu Ser Gly Arg Val Glu Phe Gly Ser Tyr Ala Asp Ala Leu Trp
319      290      295      300
321 Trp Gly Val Val Thr Val Thr Thr Ile Gly Tyr Gly Asp Lys Val Pro
322 305      310      315      320
324 Gln Thr Trp Val Gly Lys Thr Ile Ala Ser Cys Phe Ser Val Phe Ala
325      325      330      335
327 Ile Ser Phe Phe Ala Leu Pro Ala Gly Ile Leu Gly Ser Gly Phe Ala
328      340      345      350
330 Leu Lys Val Gln Gln Lys Gln Arg Gln Lys His Phe Asn Arg Gln Ile
331      355      360      365
333 Pro Ala Ala Ala Ser Leu Ile Gln Thr Ala Trp Arg Cys Tyr Ala Ala
334      370      375      380
336 Glu Asn Pro Asp Ser Ser Thr Trp Lys Ile Tyr Ile Arg Lys Ala Pro
337 385      390      395      400
339 Arg Ser His Thr Leu Leu Ser Pro Ser Pro Lys Pro Lys Lys Ser Val
340      405      410      415
342 Val Val Lys Lys Lys Lys Phe Lys Leu Asp Lys Asp Asn Gly Val Thr
343      420      425      430
345 Pro Gly Glu Lys Met Leu Thr Val Pro His Ile Thr Cys Asp Pro Pro
346      435      440      445
348 Glu Glu Arg Arg Leu Asp His Phe Ser Val Asp Gly Tyr Asp Ser Ser
349      450      455      460
351 Val Arg Lys Ser Pro Thr Leu Leu Glu Val Ser Met Pro His Phe Met

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## VERIFICATION SUMMARY

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Input Set : N:\Crf3\RULE60\09597732.txt

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L:442 M:258 W: Mandatory Feature missing, <223> not found for SEQ ID#:3  
L:442 M:341 W: (46) "n" or "Xaa" used, for SEQ ID#:3  
L:445 M:258 W: Mandatory Feature missing, <223> not found for SEQ ID#:3  
L:445 M:341 W: (46) "n" or "Xaa" used, for SEQ ID#:3  
L:447 M:258 W: Mandatory Feature missing, <223> not found for SEQ ID#:3  
L:447 M:341 W: (46) "n" or "Xaa" used, for SEQ ID#:3